

Title (en)
METHODS FOR NON-INVASIVE ASSESSMENT OF GENETIC ALTERATIONS

Title (de)
VERFAHREN ZUR NICHTINVASIVEN BEURTEILUNG GENETISCHER VERÄNDERUNGEN

Title (fr)
PROCÉDÉS D'ÉVALUATION NON INVASIVE D'ALTERATIONS GENETIQUE

Publication
EP 3571615 B1 20240124 (EN)

Application
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Abstract (en)
[origin: WO2018136888A1] Technology provided herein relates in part to methods, processes, machines and apparatuses for non-invasive assessment of genetic alterations. In particular, a method is provided for that includes obtaining a set of sequence reads. The sequence reads each include a single molecule barcode (SMB) sequence that is a non-random oligonucleotide sequence. The method further includes assigning the sequence reads to read groups according to a read group signature. The read group signature comprises an SMB sequence and a start and end position of a nucleic acid fragment from the circulating cell free sample nucleic acid. The sequence reads comprising start and end positions and an SMB sequence similar to the read group signature are assigned to a read group. The method further includes generating a consensus for each read group, and determining the presence or absence of a genetic alteration based on the consensus for each read group.

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